Genetic knowledge, opinions and self-perceived competence of non-genetic health care providers

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Beliefs and Self-Perceived Competence of General Practitioners, Gynaecologists and Paediatricians with regard to Genetic Testing

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Abstract

The objective was to investigate several aspects of the medical competence of general practitioners (GPs), gynaecologists (GYNs) and paediatricians (PEDs) by assessing their beliefs and self-perceived competence with regard to genetic testing.
A questionnaire was sent to randomly selected GPs (n=200), GYNs (n=300) and PEDs (n=265).
The response rate for GPs, GYNs and PEDs was 64%, 69% and 72%, respectively. 35% of GPs, 44% of GYNs and 54% of PEDs were likely to routinely offer a predictive test for treatable common disorders, even when others in their specialty would not do so. Approximately 20% of the physicians thought that prenatal testing for hereditary breast cancer was appropriate, and between 1/3 and 1/2 thought that for sickle cell anaemia and almost all for DMD and CF. Furthermore, 53% would personally counsel a couple about prenatal diagnosis before referring them. 19% would express their personal opinion about prenatal diagnosis and 7% would do so about pregnancy termination. Approximately 10% of the physicians would disclose information about a predictive test for HD without permission to the patient's children. The majority of physicians felt confident that they could cope with all test-related situations.
These physicians had few reservations about routinely offering predictive testing if it was not standard practice. Beliefs about prenatal testing varied widely. Most physicians perceived their competence as adequate. However, attention should be paid to the tendency towards directive counselling and the disclosure of information to third parties, because the attitudes of some physicians were not in line with current guidelines.

Introduction

The recent discovery of an increasing number of genes involved in disease etiology has led to an increase in the number of genetic tests that are available in daily clinical practice,[1] and this will result in an increasing number of patients who wish to make use of these new technologies. Disease concepts will be redefined by genomics as new predisposing risk factors become apparent. Tests can be used for prediction, diagnosis, and optimization of treatment for most common diseases.[2] Consequently, the demand for genetic testing will increase in both genetic and non-genetic health care settings. Moreover, non-genetic physicians will often be the first in line to be confronted with questions about genetics, and it they will become increasingly important in providing genetic care in the future. However, several studies have found a lack of knowledge about genetics among physicians.[3-8]

Insufficient knowledge gives rise to concerns about beliefs and competence with regard to genetic testing. Therefore, this study focused on the beliefs and competences of non-genetic physicians with regard to genetic testing. The Royal College of Physicians and Surgeons of Canada (RCPSC) has produced a competency-based framework that describes the principal generic abilities of physicians oriented to optimal health and health care outcomes.[9] Apart from medical expertise (including the application of medical knowledge), professional medical competence includes six other areas of competence: communication skills, collaborative work, management, health advocacy, scholarship and professionalism.[9] These competencies, consisting of important observable knowledge, skills and attitudes, are currently being used in the planning of medical education. With respect to competencies and skills that are specific to genetic care, attention has been paid to non-directiveness in counselling and the autonomy of individuals,[10-12] informed consent and disclosure of genetic information to third parties.[13-15] Furthermore, a wide and complex range of skills is needed for counselling of disorders that occur more frequently in ethnic minority groups. Professionals in multi-ethnic societies need cultural competence to discuss genetic ancestry, while challenging racism.[16]

The aim of this study was to investigate the beliefs of general practitioners (GPs), gynaecologists (GYNs) and paediatricians (PEDs) towards genetic testing and the disclosure of information to third parties. Furthermore, it was investigated whether these health care providers are willing to offer new genetic tests to their patients, and whether or not they consider themselves as being capable of dealing with genetic testing.

Materials and Methods

Survey instrument

To investigate the beliefs towards genetic testing in general, self-perceived competence, genetic knowledge and opinions about preconceptional cystic fibrosis (CF) carrier-screening, we used (with permission) a validated questionnaire developed in the USA. [5] [17-19] The results concerning genetic knowledge and the opinions of physicians with regard to preconceptional CF carrier-screening have been described elsewhere.[3] [20] The questionnaire was translated into Dutch and adapted to the health care system in the Netherlands where, like in a number of other (European) countries, GPs are the primary care-providers and act as gate-keepers to the health-care system.[21] Translation into Dutch was validated by back translation into English, and revised where necessary. Five GPs pre-tested the questionnaire, after which the final version was compiled. If not otherwise stated, a multiple choice or yes-no format was used.

Knowledge score

Although the results concerning genetic knowledge have been described elsewhere [3] [20], the design and results are summarized here because the knowledge score was included as a variable in the analysis for this part of the study. The questionnaire contained 26 knowledge questions, consisting of 9 fact questions and 17 concept questions. By assigning one point for each correct answer, an overall knowledge score was created, with a maximum total score of 26 points. The knowledge scores for GPs (mean 64% correct answers, 61-66% [95% CI]), GYNs (mean 75% correct answers, 73-76% [95% CI]) and PEDs (mean 81% correct answers, 79-82% [95% CI]) differed significantly ($p<0.0001$). The 5th percentile of GPs, GYNs and PEDs, respectively, was approximately 40%, 52% and 62% correct answers. There was a specific lack of knowledge about DNA testing.

Innovativeness (likelihood that physicians would offer new genetic tests)

The degree of innovativeness of the physicians was measured by asking them how likely they would be to offer a low cost, accurate test, that had just become available for predicting a future common disorder in their patients, assuming that interventions in healthy people with positive test results could improve their prognosis. The physicians were asked whether they would offer the test: 1) if no other physicians in their specialty would do so, 2) if others in their specialty would offer the test, although it was not yet standard practice, and, 3) if offering the test had become standard practice. Innovativeness was measured on a 6-point Likert scale (ranging from very unlikely to

very likely). To simplify the analysis, this was reduced to two categories: '(very) likely' and '(very) unlikely'.

Beliefs towards genetic testing

Beliefs towards genetic testing were measured by first asking the respondents, if they would a) inform a patient and obtain consent before ordering a predictive test from which more than half of the patients would benefit, or b) explain the predictive test, but not request explicit consent, or c) inform the patient only after the results of the predictive test became known. They were then asked if they would report both favourable and unfavourable results to their patients, or only unfavourable results. Questions were also asked about the appropriateness of parental use of prenatal testing for seven different diseases: sickle cell anemia, neurofibromatosis I, Duchenne muscular dystrophy, hereditary breast cancer, Alzheimer disease, CF and phenylketonuria (PKU). There were three answer categories (yes, don't know, no). The seven diseases differ in prevalence, prognosis, variability, age of onset and availabilty of a therapy.

Finally, willingness and attitude towards counselling a couple about prenatal diagnosis for CF was assesed with three questions.The first questin was a) would they personally discuss prenatal diagnosis with a pregnant CF carrier couple before (or instead of) referring them to a genetic clinic, or b) would they immediately refer the couple to a genetic clinic. Regardless of their answer, physicians were told to imagine that the couple returned to discuss the option of prenatal diagnosis. In discussing the advantages and disadvantages of prenatal diagnosis with the couple, the physicians were asked whether, they would a) express their personal opinion or advice about prenatal diagnosis, or b) refrain from expressing an opinion. Subsequently, they were asked to imagine that this couple underwent prenatal diagnosis and that the fetus appeared to be affected. They were then asked what kind of advice they would give regarding pregnancy. They could choose between the following five options: a) to continue the pregnancy because of personal moral objection to abortion, b) to continue the pregnancy because of the improved prognosis of a child with CF, c) to terminate the pregnancy because of the burden that a child with CF will be on the family, or d) to terminate the pregnancy because of the burden that a child with CF will be to society, or e) no recommendation.

Disclosure of information to third parties

After having received information about medical and genetic issues concerning Huntington disease (HD), the physicians were asked whether they would disclose information about the genetic HD status of a healthy adult to six different third parties

(spouse, children, sibs, employer, Driving Test Organization (Dutch statutory body responsible for driving tests [CBR]), and health insurance company). These were yes/no answer options.

Self-perceived competence

The physicians were asked to imagine that during the course of a visit, the issue of testing for a genetic predisposition for a particular disease was raised. They had to assume that they were highly familiar with this disease. Their perception of their own competence regarding the offer and the use of genetic testing was measured on a 6-point Likert scale (ranging from very confident to not confident at all).
Questions about sociodemographic characteristics and practice characteristics were also asked.

Sample

In 2000, the questionnaire was sent to a sample of 200 of the 7,106 GPs practising in the Netherlands,, randomly selected by the Netherlands Institute of Health Services Research (NIVEL). In 2001, the questionnaire was sent to a random sample of 300 active members of the Dutch Society of Obstetrics and Gynaecology (N=691). Only those GYNs who confirmed that they were involved in obstetrics were included in the study because it had to be likely that they would be confronted with the topics and questions from patients that were included in the questionnaire. The active members of the Netherlands Paediatric Association were divided into two groups: members of the Section on Congenital and Inherited Disorders (SCID) (N=65) and other members (N=804). All 65 members of the SCID and 200 PEDs selected at random from the remaining 804 members also received the questionnaire in 2001.

Procedures

The questionnaires were sent by mail. A brief postal reminder was sent one week later, and a second reminder, again including the questionnaire, was sent after one month. After two months the principal investigator contacted the GPs who had not responded by phone, because of their lower response rate. All respondents were offered an incentive of 20 euros.

Analysis

All analyses were performed in SPSS 9.0 for Windows.
To identify variables associated with the innovativeness and beliefstowards genetic testing, differences between the specialties, gender, age-groups(<45 years vs. ≥ 45 years), years of experience, genetic knowledge score, type of hospital (general vs.

university), SCID membership and innovativeness were assessed with the Mann-Whitney U-test for ordinal data, the Chi-square test for categorical data and the t-test for continuous data. Differences between the mean knowledge scores and the various opinions about prenetal testing, as well as between the mean years of experience and the various opinions about prenatal testing were assessed with one-way ANOVA, with post-hoc Bonferroni correction for multiple comparisons. To adjust for covariance, variables with a p-value < 0.1 in univariate analysis were included in a multivariate logistic regression analysis. The 6-point Likert scale (ranging from very confident to not confident at all) for self-perceived competence was recalculated to obtain a total confidence score by summing up responses to the individual items. Rotated varimax factor analysis was used to extract the different components in these scale. All items loaded on one of the two factors: coping with emotional problems[a] and coping with information supply[b] Estimated with Cronbach's alpha, reliability for the whole scale and for factors [a] and [b] separately, was 0.84, 0.78 and 0.74, respectively. These confidence scales were dichotomised according to the frequency distributions of the respective score, with the 50th percentile falling in the upper group. Overall, a p-value of less than 0.05 was considered to be statistically significant (two-sided).

Results

Response rate and demographic characteristics

In the groups of 200 GPs, 300 GYNs and 265 PEDs, respectively, 5, 5 and 18 physicians were no longer practicing, and were excluded, as were another 10 GYNs who were no longer involved in obstetrics. The response rate was 64% (124/195) for the GPs, 69% (198/285) for the GYNs, and 72% (177/247) for the PEDs. The response rate for the PEDs who were SCID members was 69% (36/52) and for the other PEDs it was 72% (141/195). Only questionnaires that were completed for 75% or more were included in the analysis.

Further analyses were therefore based on the remaining 124 GPs, 197 GYNs and 176 PEDs.

Table 1 shows the sociodemographic characteristics of these physicians. Mean age and number of years of experience were within the expected range, taking into account the year of graduation. Most GPs (86%) were not active in obstetrics, and a minority of the GYNs and PEDs were working in university hospitals. NIVEL compared the 200 GPs in the sample to the entire population of GPs with regard to gender, age and practice characteristics, and the sample proved to be representative. No such information was available for the entire population of GYNs and PEDs, and therefore no such comparison could be made. No significant differences were found between the two

groups of PEDs (SCID members vs. all other members) with regard to sociodemographic characteristics, innovativeness, beliefs towards genetic testing and self-perceived competence. Therefore, the groups were combined for further analysis (Total N=176).

Table 1: Sociodemographic characteristics of the physicians

	GPs N=124	GYNs N=197	PEDs N=176
Male (%)	81	75	60
Age 40-54 years (%)	69	71	63
Year of graduation as an MD (mean)	1980	1979	1981
Years of experience as a specialist (mean)	16	13	13
Working in a university hospital (%)	-	15	37

Innovativeness

1. Offering a non-standard practice test when others would not do so. The majority of the GPs (65%), more than half of the GYNs (56%) and nearly half of the PEDs (46%) felt that it would be (very) unlikely that they would offer a new predictive test if others in their specialty did not so (Table 2). These differences between the three specialties were statistically significant (p=0.006). GYNs and PEDs working in a university hospital (58%) were significantly (p=0.02) more likely to offer such a test if others in their specialty did not so, compared to those working in a general hospital (46%). Older (≥ 45 years) physicians (52%) were also significantly (p=0.002) more likely to do so even though others in their specialty did not, compared to their younger colleagues (38%). This is consistent with the finding that more experienced (mean 16.4 years) physicians were significantly more likely to offer a test even if others in their specialty did not, compared to the those with less experience (mean 13.7 years) (p=0.02).
2. Offering a non-standard practice test, knowing that others would also do so. The majority of the GYNs and PEDs (70% and 78%, respectively) and more than half of the GPs (62%) stated that it was likely that they would offer a predictive test if others in their specialty would also do so, even though this was not standard practice. The difference between the three specialties in this respect was significant (p=0.01). Furthermore, GYNs and PEDs working in a university hospital (83%) were significantly (p=0.02) more likely to do so under these circumstances, compared to those working in a general hospital (70%).
3. Offering a standard practice test. If offering the test was standard practice, most physicians (94%, 96% and 96% for GPs, GYNs and PEDs, respectively) would be (very) likely that to offer the test.

4. In summary. New predictive genetic tests might be offered by 35-54% of physicians if others in their specialty did not, by 62-78% if others did, and by 94-96% if it was standard practice. Overall, PEDs were the most innovative in offering the tests, and GPs were most reluctant.

Table 2: Innovativeness: self-reported likelihood that physicians would offer new genetic tests to their patients under three different circumstances.

Is it likely that you would offer a low cost, accurate test, that just became available for predicting common disorder in your patients, knowing that interventions in healthy people with positive test results can improve their prognosis?	Probability of offering a genetic test *	Physician		
		GPs N= 124 (%)	GYNs N=197 (%)	PEDs N=176 (%)
When no other physicians in your specialty offer the test?	(Very) Likely	35	44	54
	(Very) Unlikely	65	56	46
When a number of physicians in your specialty offer the test, but it is not yet standard practice?	(Very) Likely	62	70	78
	(Very) Unlikely	38	30	22
After it became standard practice?	(Very) Likely	94	96	96
	(Very) Unlikely	6	4	4

* The six answer categories ranged from very likely to very unlikely. The 6-point scale was reduced to two categories: (very) likely and (very) unlikely to simplify the interpretation of the answers.

Beliefs towards genetic testing

1. Informed consent and reporting of test results. Almost all physicians (99-100%) would inform a patient and obtain consent before ordering a predictive test which is known to be of benefit to more than half of the patients would. Furthermore, almost all physicians (98-99%) would report both favourable and unfavourable results to their patients.
2. Parental request for prenatal testing. The beliefs towards the parental use of prenatal testing for different diseases showed more variation (Table 3). The test results are discussed for each disease separately, describing the significant differences per tested variable only.

Prenatal testing for Duchenne muscular dystrophy

Significantly ($p=0.02$) less GPs (89%) than GYNs (97%) and PEDs (95%) thought that use prenatal testing by parents for Duchenne muscular dystrophy was appropriate; 4%

of the GPs, 0% of the GYNs and 2% of the PEDs thought that this was inappropriate. Significantly (p=0.02) more female physicians (99%) than male physicians (93%) thought that the use of prenatal testing by parents for this disorder was appropriate. Physicians who were in favour of the test had a significantly (p< 0.001) higher knowledge score (19.4 points) than those who answered "don't know" (16.7 points).

Prenatal testing for CF

Similar results were found for CF; 83% of GPs, compared to 93% and 91% of the GYNs and PEDs, respectively, thought that the use of prenatal testing by parents for CF was appropriate (p=0.004). Only 7% of the GPs, and 1% of the GYNs and the PEDs thought that it was inappropriate. Those in favour of testing also had a significantly (p=0.02) higher knowledge score (19.4 points) than those who answered "don't know" (17.8 points).

Prenatal testing for PKU

More GYNs and PEDs working in a general hospital (64%) thought that the use of prenatal testing by parents for PKU was appropriate, compared to those working in a university hospital (48%;p=0.02); respectively 29% and 34% of the physicians working in a general and a university hospital were against testing.

Prenatal testing for neurofibromatosis

Respectively 49%, 58% and 50% of the GPs, GYNs and PEDs, thought that the use of prenatal testing by parents for Neurofibromatosis I was appropriate. No significant associations were found between the sociodemographic variables, innovativeness, knowledge scores and the attitudes towards prenatal testing.

Prenatal testing for sickle cell anemia

The results for sickle cell anemia are very different from those for Duchenne muscular dystrophy, cystic fibrosis and PKU. Significantly more (p=0.02) innovative physicians thought that the use of prenatal testing by parents for sickle cell anemia (48%) was appropriate, compared to the other physicians (35%); respectively 29% of the more innovative physcians and 37% of the non-innovative physicians thought that it was inappropriate.

Prenatal testing for hereditary breast cancer

Female physicians (59%) differed significantly (p=0.02) from male physicians (47%) in their approval of the use of prenatal testing by parents for hereditary breast cancer; 16% of the females and 27% of the males thought that it was appropriate. Significantly

(p=0.002) more younger (<45 years) physicians (56% vs 45%) thought that it was inappropriate; 16% of the younger physicians and 30% of the older physicians were in favour of testing. Furthermore, significantly (p=0.04) more GYNs and PEDs working in a university hospital (63%), compared to those working in a general hospital (47%), thought that it was inappropriate; respectively 13% and 26% of the physicians working in university and general hospitals thought that it was appropriate. Physicians who thought that it was appropriate had a significantly (p=0.01) lower knowledge score (18.6 points) than those who were against it (19.8 points) and those who answered "don't know" (18.8 points) (p=0.02).

Prenatal testing for Alzheimer disease

Significantly (p=0.02) more GYNs and PEDs working in a university hospital (66%) than GYNs and PEDs working in a general hospital (51%) thought that for the use of prenatal testing by parents for Alzheimer disease was inappropriate; whereas respectively 8% and 21% of the physicians working in a university and a general hospital thought that it was appropriate. Furthermore, physicians who thought it was appropriate had a significantly (p=0.01) lower knowledge score (18.4 points) than those who were against it (19.7 points). No other significant associations were found between the sociodemographic variables, innovativeness and knowledge scores, on the one hand, and the attitudes towrds the use of prenatal testing by parents for different diseases on the other hand.

Counselling about prenatal diagnosis and non-directiveness

Of the physicians, 53% would personally counsel a couple about prenatal diagnosis themselves before or instead of referring them to a genetic clinic. Significantly (p=0.03) less GPs (43%) than GYNs (54%) and PEDs (59%) would do so. Significantly (p=0.01) more younger (59%) than older physicians (47%) would counsel personally before referral. Physicians who would counsel before referral had significantly (p=0.01) less experience in their field of practice (13.4 years) and a significantly (p<0.0001) higher knowledge score (19.9 points) than physicians who would refer immediately (16.8 years; 18.6 points). In multivariate analysis it was found that a lower level of knowledge was significantly associated with immediate referral (odds 0.9; CI 0.86-0.99). Nineteen percent of the physicians would express their personal opinion about prenatal diagnosis during the consultation (24% of GPs, 15% of GYNs, 20% of PEDs). Significantly (p=0.004) more older (24%) than younger physicians (13%) and significantly (p < 0.001) more males (25%) than females (7%) would do so. Those who would express their personal opinion about prenatal diagnosis were significantly (p=0.00) more experienced in their field (18.7 years) than those who would not (13.9

years). Male gender was found to be significantly associated with expressing a personal opinion in multivariate analysis (f:m odds 0.3; CI 0.1-0.6). Only 7% of the physicians (10% of GPs, 3% of GYNs, 8% of PEDs) would express their own opinion about pregnancy termination. The difference between the three specialties was statistically significant (p=0.046). Significantly (p=0.02) more older (9%) than younger physicians (4%) would express their opinion about pregnancy termination. Those who would do so were significantly (p=0.03) more experienced in their field (20.1 years) and had a significantly (p=0.01) lower knowledge score (17.5 points) than those who would not (14.6 years; 19.4 points). Furthermore, significantly (p=0.001) more males (9%) than females (1%) would express their opinion about pregnancy termination. In multivariate analysis, male gender was again significantly associated with directive counselling (f:m odds 0.1; CI 0.0-0.6).

Table 3: Attitudes of the physicians towards prenatal testing by parents for different diseases

Disease	Appropriateness of prenatal testing	Physician		
		GPs N=124 (%)	GYNs N=197 (%)	PEDs N=176 (%)
Duchenne muscular dystrophy	Yes	89	97	95
	Don't know	7	3	3
	No	4	0	2
Cystic fibrosis	Yes	83	93	91
	Don't know	10	6	7
	No	7	1	1
Phenylketonuria	Yes	62	60	61
	Don't know	13	12	8
	No	25	28	31
Neurofibromatosis I	Yes	49	58	50
	Don't know	30	27	24
	No	21	15	26
Sickle cell anemia	Yes	34	39	47
	Don't know	33	25	22
	No	33	36	31
Hereditary breast cancer	Yes	26	27	17
	Don't know	27	25	29
	No	47	48	54
Alzheimer disease	Yes	17	21	14
	Don't know	23	28	28
	No	60	51	58

Disclosure of information to third parties

Very few physicians thought that information about a predictive test for Huntington disease should be disclosed to a spouse (2-4%) or sibs (2-3%) without permission from the person tested. Approximately 10% of the physicians (8-11%) would disclose the information without permission to the patient's children. None of the physicians would inform the employer. Approximately 3% of the GPs and less than 1% of the GYNs and PEDs would inform the CBR and only 2% of the PEDs and none of the GPs or GYNs intended would inform the health insurance company.

Self-perceived competence

Table 4 shows how confident the physicians felt in their ability to perform various different actions related to genetic testing. Most of them were (moderately) confident about their actions in all kinds of test-related situations. Two components could be extracted: coping with emotional problems (a) which consisted of 5 items (e.g. helping a patient to cope with an unfavourable test result), and coping with information supply (b) consisting of 4 items (e.g. referring the patient to an appropriate specialist) (Table 4). GPs (67%) felt significantly ($p<0.0001$) less confident than PEDs (42%), and GPs (73%) also felt significantly ($p<0.0001$) less confident than PEDs (31%) in coping with information supply. GYNs and PEDs working in a general hospital (55%) felt significantly ($p<0.0001$) less confident than GYNs and PEDs working in a university hospital (29%). This applies to coping with emotional problems (48% vs 32%) as well as coping with information supply (49% vs. 21%). GPs (73%) felt significantly ($p<0.0001$) less confident than PEDs (31%) in coping with information supply. Those who felt more confident had a significantly ($p<0.0001$) higher knowledge score (19.9) than the other physicians (18.6), and those who perceived themselves to be more confident in coping with information supply had significantly ($p<0.01$) higher knowledge scores (20.1) than the other physicians (18.3). Gender, age and years of experience did not influence their perceived competence.

Table 4: Self-perceived competence of the physicians

Ability	Competence *	Physician		
		GPs N= 124 (%)	GYNs N=197 (%)	PEDs N=176 (%)
To help a patient to understand the risk of developing a genetic disease[b]	(Moderately) Confident	79	86	86
	(Moderately) Not Confident	18	11	7
To discuss with a patient fears and concerns about getting a genetic disease[a]	(Moderately) Confident	92	86	83
	(Moderately) Not Confident	3	8	7
To discuss with a patient risks and benefits of treatment for the genetic disease[b]	(Moderately) Confident	85	84	86
	(Moderately) Not Confident	12	10	4
To help a patient to decide whether or not to be genetically tested[a]	(Moderately) Confident	80	83	88
	(Moderately) Not Confident	16	15	9
To obtain additional relevant information about a test from a colleague or from books/journals[b]	(Moderately) Confident	71	80	76
	(Moderately) Not Confident	25	8	4
To discuss the meaning and implications for a patient of a negative (favourable) test[a]	(Moderately) Confident	87	91	86
	(Moderately) Confident	5	4	7
To help a patient to understand the implications of a positive (unfavourable) test result[b]	(Moderately) Confident	86	89	87
	(Moderately) Not Confident	14	7	8
To help a patient to cope with a positive (unfavourable) test result[a]	(Moderately) Confident	91	85	86
	(Moderately) Not Confident	2	12	11
To refer the patient to an appropriate specialist[b]	(Moderately) Confident	75	70	67
	(Moderately) Not Confident	2	1	1

* The answer categories (1-6) ranged from very confident to not confident at all. Only the answer categories confident/ moderately confident and moderately not confident/ not confident are shown.

Two components could be extracted with factor analysis: coping with emotional problems [a] and coping with information supply [b]

Discussion

The objective of this study was to investigate several aspects of the medical competence of GPs, GYNs and PEDs by assessing their beliefs and self-perceived competence with regard to genetic testing. Specific attention was paid to non-directiveness in counselling, the autonomy of individuals, informed consent and disclosure of genetic information to third parties, because it can be argued that these issues should be regarded as important for "best clinical practice".[10-15] As many as 46-65% of these physicians had reservations about offering genetic testing, if it was not standard practice. Overall, the PEDs were the most willing to offering genetic testing, and GPs were the most reluctant. Beliefs towards prenatal testing varied widely for different diseases. Most physicians perceived their competence as adequate. These findings are in line with similar or even higher percentages of reluctance reported in other studies. [22] However, physicians seem to be prepared to provide genetic care in the future in the same way as they already apply various guidelines in other domains of healthcare.

An important factor influencing the implementation of genetic tests by non-genetic health care providers is their innovativeness.[23] In our study, the minority of the GPs (35%), less than half of the GYNs (44%), and approximately half of the PEDs (54%) felt that it would be (very) likely that they would offer a new predictive test if others in their specialty would not. This is in line with the findings of a study carried out in the USA, in which 51% of the physicians stated that they would do so[5], but in our study there was no difference in the knowledge scores of innovative and non-innovative physicians as was found in the USA study. [5]

As nearly all the physicians in our study would inform a patient and obtain consent before ordering a predictive test, and would report both favourable and unfavourable results to their patients, there should be no problems in this respect in the provision of genetic tests by these non-genetic health care providers.

The beliefs of the physicians with regard to the use of prenatal testing by parents for different diseases varied widely. However, all the answers concerning the appropriateness of the use of prenatal diagnosis by parents ranged in the same way from high (Duchenne muscular dystrophy) to low acceptance (Alzheimer disease). This is probably related to the severity of the disease, and possibly also to individual knowledge about these diseases. Most physicians were in favour of the use of prenatal diagnosis for Duchenne muscular dystrophy and CF. Physicans who belong to specialties with exposure to these genetic disorders and who had more knowledge about genetics were more likely to consider this as appropriate. We reported earlier that most GYNs and PEDs are aware of the availability of a DNA test for CF, which is in

line with the findings of a UK study. [3 24] However, only two-thirds of GPs are aware of the availability of a CF test.[3] Lack of knowledge about CF-testing has already been shown to be a problem, misleading people planning a pregnancy who ask questions about CF-testing, but are not receiving the correct advice from their physicians.[25] Approximately 60% of the physicians thought that the use of prenatal testing by parents for PKU was appropriate. That is a remarkably high percentage because of the availability of treatment for children with PKU and its widespread inclusion in neonatal screening programmes. Approximately half of the physicians (49-58%) were in favour of the use of prenatal testing by parents for neurofibromatosis I. This difference of opinion could be due to the variance in phenotype. Approximately one third of the physicians (31-36%) thought that the use of prenatal testing by parents for sickle cell anemia was inappropriate, perhaps because they underestimated the severity of the disease. This is worriying because, according to Petrou, most sickle cell carrier couples, if referred in the first trimester, request prenatal diagnosis.[26] In our study in the Netherlands, approximately three quarters of the GPs and half of the GYNs and PEDs were unaware of the availability of a DNA test for sickle-cell anaemia.[3] Early counselling of these couples during, or preferably even before pregnancy, is therefore difficult to achieve. Our results might well illustrate a lack of cultural competence and awareness.

Fewer physicians were in favour of the use of prenatal diagnosis by parents for late onset diseases such as hereditary breast cancer and Alzheimer disease. Approximately 50% (47-54%) of the physicians thought that the use prenatal diagnosis for hereditary breast cancer was not appropriate, although it has been argued by Dutch clinical geneticists that it is acceptable on request after extensive counselling.[27]

More than half of the respondents (53%) would counsel couples personally before or instead of referral to a genetic clinic. This is in line with the findings of a study in the USA, in which 49% of the respondents stated that they would counsel personally about prenatal diagnosis.[18] In that study as well as in our study, younger physicians and physicians with a higher knowledge score were more likely to counsel than older physicians.[18] Only 19% of the physicians in our study would express their personal opinion about prenatal diagnosis, compared with 44% in the USA study.[18] In both studies males were more likely than females to express their personal opinion. In our study, 7% of the physicians would express an opinion about pregnancy termination, which is comparable to 10% in the USA study.[18] In both studies, GYNs were more reluctant than GPs to express an opinion.[18] Another study in the Netherlands also found that GPs supported a directive method of counselling.[28] Williams et al. argued that the issue of non-directiveness may be undermined by a tight time schedule and less familiarity with this principle among midwives and obstetricians, who increasingly

carry out genetic counseling during pregnancy, instead of specialized genetic clinics.[29] Hall et al. found a positive association between the amount of negative information parents are given about a sex chromosome anomaly and the decision to terminate the affected pregnancy.[30] Marteau et al. also reported that the chance that parents would terminate a pregnancy for a sex chromomal anomaly was smaller when counselled by a clinical geneticist only than when counselled by a obstetrician, paediatrician or midwife.[31]

Very few physicians would disclose Huntington test results to non-family members (0-3%). This is in line with findings in the USA, where only 1 – 5% of the respondents would do so.[32] Also, only relatively few (2-11%) physicians in our study would disclose information about this predictive genetic test to family members, especially the patient's children, without permission from the patient. This percentage is much lower than in the USA study, in which 16-29% of respondents would do so (29%).[32] The health risk of children is influenced by this information, but it is not clear that disclosure of this information is appropriate. Disclosure of confidential information to relatives is only acceptable if it will prevent serious imminent harm to a person.[13-15] Physicians might, however, be of the opinion that children have the right to know their own health risks.

Most of the physicians in our study felt (moderately) confident about their ability to cope in all genetic test-related situations mentioned in this study. This is not surprising, because a number of these abilities apply to every day health care practice. However, in spite of their own perception of adequate competence, more training is important. Special attention should be paid to improve the lack of genetic knowledge and the awareness of genetic tests, the directive way of counselling (specifically for prenatal diagnosis and pregnancy termination) and the disclosure of information to third parties, as stated above. Fortunately, the physicians who were more confident had higher knowledge scores than the others. Cardiologists have been reported to consider their knowledge and practical genetic skills as insufficient, and to have stressed the need for more education.[33] This is in line with the findings of other studies, in which it has been shown that non-genetic health care providers feel a need for more education in genetics.[8 34-36] One could argue that some of the physicians in our study were well aware of their own lack of genetic knowledge, because those with a lower knowledge score felt less confident about their own abilities than the others. However, in general, the physicians felt confident and the majority intend to offer predictive genetic tests. Some of the gold standards for non-directiveness and confidentiallity were not adhesed to by all respondents.

Limitations of the study

This study only reports data on self-reported beliefs and competence, and not on actual abilities. However, these aspects were measured in large samples of physicians, with sufficient response rates, and therefore the results should reflect their actual abilities. Secondly, in some countries GPs do not have the role of "gate-keeper", due to differences in the health care systems, and therefore the results of this study can only be extrapolated with caution to the situation in other countries.

Conclusion

In general, GPs, GYNs and PEDs are willing to counsel a patient about genetic testing, would offer a predictive test if it is common practice and perceive their competence as adequate. However, in medical education attention should be paid to the lack of genetic knowledge, the tendency towards directive counselling and (even in a minority) the disclosure of genetic information to third parties.[3 37] There seem to be few thresholds in the implementation of genetic testing by non-genetic health care professionals. However, since some "gold standards" for professional clinical genetics are not adhered to by all GPs, GYNs and PEDs, further training is needed in order to optimize the benefits of genetic testing for patients in the future.

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